# OTC gene

ornithine carbamoyltransferase

## Normal Function

The *OTC* gene provides instructions for making the enzyme ornithine transcarbamylase. This enzyme participates in the urea cycle, a series of reactions that occurs in liver cells. The urea cycle processes excess nitrogen, generated when protein is used by the body, into a compound called urea that is excreted by the kidneys. Excreting the excess nitrogen prevents it from accumulating in the form of ammonia, which is toxic, especially to the nervous system.

The specific role of the ornithine transcarbamylase enzyme is to control the reaction in which two compounds, carbamoyl phosphate and ornithine, form a new compound called citrulline.

## Health Conditions Related to Genetic Changes

ornithine transcarbamylase deficiency

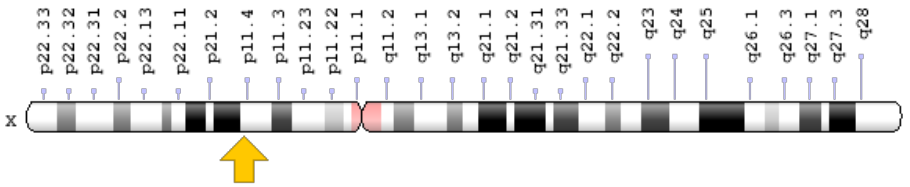
More than 500 *OTC* gene mutations have been identified in people with ornithine transcarbamylase deficiency, an inherited disorder that causes ammonia to accumulate in the blood. Ammonia, which is formed when proteins are broken down in the body, is toxic if the levels become too high. The nervous system is especially sensitive to the effects of excess ammonia.

The *OTC* gene mutations that cause ornithine transcarbamylase deficiency result in an ornithine transcarbamylase enzyme that is shorter than normal or the wrong shape, or prevent any enzyme from being produced. The shape of an enzyme affects its ability to control a chemical reaction. If the ornithine transcarbamylase enzyme is misshapen or missing, it cannot fulfill its role in the urea cycle. Excess nitrogen is not converted to urea for excretion, and ammonia accumulates in the body. Accumulation of ammonia causes neurological problems and other signs and symptoms of ornithine transcarbamylase deficiency.

## Chromosomal Location

Cytogenetic Location: Xp11.4, which is the short (p) arm of the X chromosome at position 11.4

Molecular Location: base pairs 38,352,483 to 38,421,450 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- MGC129967
- MGC129968
- OCTD
- ornithine carbamoyltransferase precursor
- ornithine transcarbamylase
- OTC_HUMAN

## Additional Information & Resources

Educational Resources

- Biochenmistry (fifth edition, 2002): Ammonium Ion Is Converted Into Urea in Most Terrestrial Vertebrates
  https://www.ncbi.nlm.nih.gov/books/NBK22450/

GeneReviews

- Ornithine Transcarbamylase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK154378
- Urea Cycle Disorders Overview
  https://www.ncbi.nlm.nih.gov/books/NBK1217

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28OTC%5BTIAB%5D%29+OR+%28ornithine+carbamoyltransferase%5BTIAB%5D%29%29+AND+%28%28carbamoyl-phosphate:l-ornithine+carbamoyltransferase%5BMAJR%5D%29+OR+%28ornithine+transcarbamylase%5BMAJR%5D%29+OR+%28ornithine+carbamylphosphate+transferase%5BMAJR%5D%29+OR+%28carbamoyltransferase,+ornithine%5BMAJR%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- ORNITHINE CARBAMOYLTRANSFERASE
  http://omim.org/entry/300461

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_OTC.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=OTC%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8512
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5009
- UniProt
  http://www.uniprot.org/uniprot/P00480

## Sources for This Summary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  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/25958381
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  https://www.ncbi.nlm.nih.gov/books/NBK1217
- Helman G, Pacheco-Colón I, Gropman AL. The urea cycle disorders. Semin Neurol. 2014 Jul;34(3): 341-9. doi: 10.1055/s-0034-1386771.
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  http://omim.org/entry/300461


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